HINT1 gene

histidine triad nucleotide binding protein 1

Normal Function

The *HINT1* gene provides instructions for making a protein called histidine triad nucleotide-binding protein 1 (HINT1). The function of this protein is not clear. Laboratory studies show that the HINT1 protein has the ability to attach (bind) to certain types of molecules called nucleotides. HINT1 breaks down particular nucleotides through a process called hydrolysis. However, it is not known if the HINT1 protein performs the same reaction in the body or what effects it has.

Although the mechanisms are not completely understood, the HINT1 protein is involved in the nervous system. In nerve cells (neurons), HINT1 binds to signaling proteins called receptors that relay signals affecting nervous system function. HINT1 appears to stabilize the interaction of different receptors and regulate the effects of their signaling.

The HINT1 protein is also involved in programmed cell death (apoptosis), which occurs when cells are no longer needed. In addition, by blocking the activity of certain genes, HINT1 acts as a tumor suppressor, which means that it keeps cells from growing and dividing too rapidly or in an uncontrolled way.

Health Conditions Related to Genetic Changes

autosomal recessive axonal neuropathy with neuromyotonia

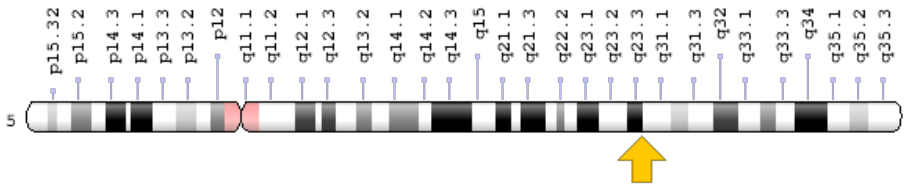
At least nine mutations in the *HINT1* gene have been found to cause autosomal recessive axonal neuropathy with neuromyotonia. This neurological condition affects the peripheral nerves, which connect the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch. People with this condition typically have muscle weakness in the feet, legs, and hands and delayed relaxation of muscles after tensing (neuromyotonia). Some affected individuals have mildly impaired sensations.

The *HINT1* gene mutations that cause this condition change single protein building blocks (amino acids) in the HINT1 protein. These changes reduce or eliminate the protein's ability to perform the hydrolysis reaction. Sometimes the altered protein is broken down prematurely. It is not clear how loss of functional HINT1 protein affects the peripheral nerves or leads to the signs and symptoms of this condition.

Chromosomal Location

Cytogenetic Location: 5q23.3, which is the long (q) arm of chromosome 5 at position 23.3

Molecular Location: base pairs 131,159,283 to 131,165,348 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- adenosine 5'-monophosphoramidase
- HINT
- histidine triad nucleotide-binding protein 1
- NMAN
- PKCI-1
- PRKCNH1
- protein kinase C inhibitor 1
- protein kinase C-interacting protein 1

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HINT1%5BTIAB%5D%29+OR+%28histidine+triad+nucleotide+binding+protein+1%5BTIAB%5D%29%29+OR+%28%28histidine+triad+nucleotide-binding+protein+1%5BTIAB%5D%29+OR+%28NMAN%5BTIAB%5D%29+OR+%28PKCI-1%5BTIAB%5D%29+OR+%28PRKCNH1%5BTIAB%5D%29+OR+%28protein+kinase+C+inhibitor+1%5BTIAB%5D%29+OR+%28protein+kinase+C-interacting+protein+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- HISTIDINE TRIAD NUCLEOTIDE-BINDING PROTEIN 1
  http://omim.org/entry/601314

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HINT1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HINT1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4912
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3094
- UniProt
  http://www.uniprot.org/uniprot/P49773

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